

Application No. 10/672,937
Amendment and Reply Dated January 10, 2008
In Response to Office Action Dated October 19, 2007

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Amendments to the Claims

This listing of claims will replace all prior versions, and listings, of claims in the application:

Listing of claims:

1. (Currently amended) ~~An automated~~ A method for sequence evaluation used to compare outputting variants between sample sequence information relating to at least one sample ~~against and~~ reference sequence information relating to at least one reference, the method comprising:

acquiring sample sequence information relating to the at least one sample ~~and to the at least one reference;~~

assembling consensus sequence information from the sample sequence information;

generating rule-based criteria, the rule-based criteria comprising pre-selected rules for at least one of scan position differences, peak height ratios, peak area ratios, and base composition;

~~evaluating the sequence information relating to the at least one sample to identify~~

identifying ambiguous bases present within the sample consensus sequence information by applying a comparing the consensus sequence information to the rule-based criteria, wherein ambiguous bases are distinguished from unambiguous bases on the basis of that do not meet the pre-selected rules for at least one of the following rule-based criteria: (a) scan position differences, (b) peak height ratios, (c) peak area ratios, and (d) base composition are labeled ambiguous;

determining what percentage of the bases present within the consensus sequence information are ambiguous bases;

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trimming the consensus sequence information until the percentage of ambiguous bases is below a user-defined threshold to form trimmed consensus sequence information;

~~evaluating the quality and coverage of the sample sequence information in comparison to the reference sequence information to identify reportable ranges and sequence variants for the sample sequence information based on a quality value determined at least by the identified ambiguous bases and unambiguous bases, wherein the reportable ranges comprise base sequence ranges related to the at least one sample having an associated quality value exceeding a confidence threshold, and being usable for comparison matching; and~~

determining variant information between the trimmed consensus sequence information and the reference sequence information; and

~~outputting at least the reportable ranges and sequence variants~~ the variant information to a file user.

2. (Currently amended) The method of claim 1, wherein the generated rule-based criteria for assessing comprises pre-selected rules for scan position differences to differentiate between ~~ambiguous and unambiguous bases further~~ and the method further comprises identifying differences between scan positions of major and minor peaks, within the sample sequence information, which fall below an empirical threshold.

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3. (Original) The method of claim 2, wherein the empirical threshold associated with identifying differences between scan positions is in the range of approximately 0 to approximately 3.
4. (Currently amended) The method of claim 1, wherein the generated rule-based criteria ~~for assessing comprises pre-selected rules for scan position differences for differentiating between ambiguous and unambiguous bases further~~ and the method further comprises identifying differences between scan positions of major and minor peaks within the sample sequence information, which reside above, below, or are substantially equivalent to, a user-defined threshold.
5. (Currently amended) The method of claim 1, wherein the generated rule-based criteria ~~for assessing comprises pre-selected rules for peak height ratios to differentiate between ambiguous and unambiguous bases further~~ and the method further comprises assessing identifying differences between peak height ratios for major and minor peaks, within the sample sequence information, which exceed an empirical threshold.
6. (Currently Amended) The method of claim 5, wherein the empirical threshold associated with ~~assessing~~ the peak height ratios is in the range of approximately 0.3 to approximately 1.0.
7. (Currently amended) The method of claim 1, wherein the generated rule-based criteria ~~for assessing comprises pre-selected rules for peak height area ratios for differentiating between~~

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~~ambiguous and unambiguous bases further~~ and the method further comprises ~~assessing~~ identifying ~~differences between~~ peak area ratios of major and minor peaks₂ within the sample sequence information₂ which reside above, below, or are substantially equivalent to₂ a user-defined threshold.

8. (Currently amended) The method of claim 1, wherein the generated rule-based criteria for ~~assessing~~ comprises pre-selected rules for peak area ratios ~~to differentiate between ambiguous and unambiguous bases further~~ and the method further comprises ~~assessing~~ identifying differences ~~between~~ peak area ratios for major and minor peaks₂ within the sample sequence information₂ which exceed an empirical threshold.

9. (Currently amended) The method of claim 8, wherein the empirical threshold associated with ~~assessing~~ the peak area ratios is in the range of approximately 0.3 to approximately 1.0.

10. (Currently amended) The method of claim 1, wherein the generated rule-based criteria ~~for assessing~~ comprises pre-selected rules for peak area ratios ~~for differentiating between ambiguous and unambiguous bases further~~ and the method further comprises ~~assessing~~ identifying differences ~~between~~ peak area ratios of major and minor peaks₂ within the sample sequence information₂ which reside above, below, or are substantially equivalent to₂ a user-defined threshold.

11. (Currently amended) The method of claim 1, wherein the generated rule-based criteria ~~for assessing~~ comprises pre-selected rules for base composition ~~to differentiate between ambiguous and~~

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~~unambiguous bases further~~ and the method further comprises determining major and minor peaks within the sample sequence information and determining if the major and minor peaks within the sample sequence information are both purines or both pyrimidines.

12. (Canceled).

13. (Currently amended) The method of claim 1, wherein the generated rule-based criteria for ~~distinguishing between ambiguous and unambiguous bases~~ comprises pre-selected rules for identifying consecutive runs of bases exceeding an empirical threshold.

14. (Currently Amended) The method of claim 13, wherein the empirical threshold ~~associated with~~ comprises identifying for comparing consecutive runs of bases is in the range of approximately 10 to approximately 13.

15. (Currently amended) The method of claim 1, wherein the generated rule-based criteria for ~~distinguishing between ambiguous and unambiguous bases identifies~~ further comprises identifying consecutive runs of bases which reside above, below, or are substantially equivalent to a user-defined threshold.

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16. (Currently amended) The method of claim 1, ~~wherein~~ further comprising:
excluding identified ambiguous bases ~~are excluded~~ from the evaluation of quality and
coverage of the sample sequence information and assembling exclusionary consensus sequence
information; and
determining variant information between the exclusionary consensus sequence information
and the reference sequence information.

17. (Canceled)

18. (Currently Amended) The method of claim 1, wherein the sample sequence information relating to the at least one sample and the reference sequence information relating to the at least one reference ~~comprises~~ comprise mitochondrial DNA sequence information.

19-44. (Canceled)

45. (New) The method of claim 1, wherein the identifying ambiguous bases further comprises assigning a quality value to those bases that have been identified as ambiguous.

46. (New) The method of claim 1, wherein trimming the consensus sequence information comprises trimming the edges of the consensus sequence information.

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47. (New) A computer readable medium having stored thereon instructions which cause a general purpose computer to perform the steps of:

- acquiring sample sequence information relating to the at least one sample;
- assembling consensus sequence information from the sample sequence information;
- generating rule-based criteria, the rule-based criteria comprising pre-selected rules for at least one of scan position differences, peak height ratios, peak area ratios, and base composition;
- identifying ambiguous bases present within the consensus sequence information by comparing the consensus sequence information to the rule-based criteria, wherein bases that do not meet the pre-selected rules for at least one of the rule-based criteria are labeled ambiguous;
- trimming the consensus sequence information until the percentage of ambiguous bases is below a user-defined threshold, to form trimmed consensus sequence information;
- determining variant information between the trimmed consensus sequence information and the reference sequence information; and
- outputting the variant information to a user.